

进行性肌营养不良患者Myostatin基因的表达分析

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摘要 进行性肌营养不良是一组以进行性骨骼肌萎缩和无力为特征的肌源性肌病。肌肉抑制素 (Myostatin) 是最近发现的骨骼肌生长发育抑制因子。为探讨Myostatin基因与进行性肌营养不良病理发生的相关性, 采用RT-PCR方法克隆了患者的Myostatin基因并测序, 分析肌营养不良患者是否存在Myostatin基因突变; 然后采用半定量RT-PCR方法检测患者中Myostatin基因的表达水平是否发生改变, 同时用Western blot方法分析了肌营养不良患者中Myostatin蛋白的表达情况。结果发现, 所研究的肌营养不良患者中没有携带Myostatin基因突变, 但一些患者的Myostatin基因转录水平降低, 部分患者Myostatin蛋白加工障碍。结果提示, 一些类型 (亚型) 的进行性肌营养不良可能与肌肉抑制素Myostatin基因表达异常、蛋白加工障碍有关。

关键词 [进行性肌营养不良](#); [肌肉抑制素基因](#); [蛋白加工](#)

分类号

Altered Expression of Myostatin Gene in the Progressive Muscular Dystrophy Patients

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Abstract

Progressive muscular dystrophy is a group of inherited disorders characterized by progressive skeletal muscle wasting and weakness, which was not of neurogenic origin. Myostatin, a new member of the TGF- β super-family, is a negative regulator of skeletal muscle growth. To investigate possible involvement of myostatin in development of progressive muscular dystrophy, we cloned and sequenced myostatin cDNAs from the progressive muscular dystrophy patients by RT-PCR. Levels of myostatin mRNA and protein in the patients were analyzed by semi-quantitative RT-PCR and Western Blot method, respectively. We did not find any mutations in the myostatin cDNA sequences from progressive muscular dystrophy patients in this study. However, we found that the levels of myostatin transcripts are reduced in some patients and the processing and maturation of myostatin protein are inhibited in some patients. Our data demonstrated that the pathogenesis of some types or subtypes of progressive muscular dystrophy is probably associated with the altered myostatin expression and the processing inhibition of myostatin protein.

Key words [progressive muscular dystrophy](#) [myostatin](#) [protein processing](#)

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